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Klippel-Feil Syndrome Associated With Butterfly and Hemi Vertebrae, Bilateral Fused Ribs and Eagle Syndrome an Uncommon Association

KEYWORDS: Klippel-Feil, butterfly vertebrae, fused ribs , Eagle syndrome.

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ABSTRACT *Klippel-Feil syndrome (KFS) is a complex heterogeneous entity that results in cervical vertebral fusion. The classic clinical triad of a short neck, low posterior hairline, and restricted neck motion is considered to be present in < 50% of patients with this syndrome. We report a case of 16-years-old female presented with bony ankylosis of multiple cervical vertebrae associated with butterfly vertebrae, multiple fused ribs and bilateral calcified stylohyoid ligaments.*

CASE REPORT-

A 16-year-old female presented to our department with the history of torticollois and neck and shoulder discomfort after minor neck trauma at 10 years of age. At clinical examination markedly reduced vertebral range of motion seen with short neck and low posterior hairline. However both the scapulas were normal in position and no other associated scoliosis seen. Patient also complained of mild intermittent otalgia and dysphagia. Patient complained of pain in neck while rotating the head . CT scan was done with 16 slice CT scanner. Non contrast CT of whole spine performed , revealed multiple cervical block vertebrae, (C5 to T1 vertebrae) , multiple vertebrae showed midline sagittal cleft with tapered margins of defect suggested butterfly vertebra (C1,C4 and T1) , hemivertebra (C7) within the block vertebrae(Fig 1 and 2). A dextroconvex scoliosis seen in cervical spine. Bilateral , multiple fused (left upper three ribs and right upper two ribs) seen. Calcification of bilateral stylohyoid ligament seen which was symptomatic suggested Eagle syndrome(Fig 2). Abdominal ultrasound was done which revealed no associated renal anomalies.

DISCUSSION-

Klippel-Feil syndrome is defined by congenital fusion of at least two cervical vertebrae with an absence of the intervertebral disc. Klippel-Feil syndrome is defined by congenital fusion of at least two cervical vertebrae along with an absence of the intervertebral discs. The syndrome was first reported by Maurice Klippel and Andre Feil in 1912 independently . It is a result of failure of normal segmentation and fusion processes of the cervical somites, which occur between the third and seventh week of embryogenic development [2]. The incidence of Klippel-Feil syndrome is approximately 1:42000. The syndrome is slightly more common in women (60%) than in men [2].The classical triad of the anomaly consists of a short neck, limitation of head and neck movements and a low posterior hairline. This triad is seen in upto 50% of patients [3]. Some presents with weakness, spasticity, dysmetria, nystagmus, sensory loss, and lower cranial nerve involvement due to progressive cord and brain stem compression, as minor trauma alters force transfer mechanics and make nonfused segments excessively mobile causing symptoms. Klippel-Feil can be associated with several organ anomalies such as congenital heart defects (most often ventricular septal defect), deafness, learning disabilities and renal anomalies [2]. Other skeletal deformities such as scoliosis and a Sprengel deformity (rotated and elevated scapula) are also common findings [3].Midline vertebral fusion defects such as butterfly vertebra , hemivertebra, malformed ribs , variable degree of fusion of ribs are also associated. Some rare associations such as Poland syndrome, vascular anatomical variation , mondini syndrome and amyotropic lateral sclerosis also reported.KFS has been classified into three types depending on the extent and location of vertebral fusion as well as associated vertebral abnormalities. There are three types. In type I, there are numerous fused cervical vertebrae and upper thoracic vertebrae. In type II, the patient presents with fusion of one or two vertebrae, most often C2-C3 or C5-C6. In type III, fusion of both cervical and lower thoracic or lumbar vertebrae occur. This type is often associated with the above mentioned systemic anomalies [3]. Type II is commonest; C2-3 and C5-6 inter-spaces are most often fused.Fusion of cervical and upper thoracic vertebrae seen with absence or hypoplasia of intervertebral disc. other radiographic findings include fusion of the facet joints and spinous processes. Imaging may sometimes show the wasp-waist sign .The wasp-waist sign refers to the vertebral body narrowing and associated concavity of the anterior and posterior surfaces of the vertebrae at the level of the fused segment [4] Most of the patients are asymptomatic. However, there is an increased risk of developing spontaneous or post-traumatic neurological injury. Patients may present with radiculopathy, myelopathy or quadriplegia caused by spondylolysis or instability of vertebrae adjacent to the fused segment.In the differential diagnosis of Klippel Feil Syndrome one must consider cleido cranial dysostoses, Sprengel's deformity, Wildervanck's syndrome and those abnormalities associated with webbing of the neck without bony defect like Turner's syndrome and pterigium colli. (5).

Eagle syndrome refers to symptomatic elongation of the styloid process or calcified stylohyoid ligament . It is often bilateral. Although approximately 4% of the population is thought to have an elongated styloid process, only a small percentage (between 4% and 10.3%) of this group

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is thought to actually be symptomatic (6). No data could be found to correlate degree of elongation of the styloid to severity of symptoms. Normal length of the styloid in an adult is approximately 2.5 cm whereas an elongated styloid is generally >3 cm in length .The actual cause of the elongation is a poorly understood process. Several theories have been proposed: 1) congenital elongation of the styloid process due to persistence of a cartilaginous analog of the stylohyal (one of the embryologic precursors of the styloid), 2) calcification of the stylohyoid ligament by an unknown process, and 3) growth of osseous tissue at the insertion of the stylohyoid ligament (7). However it is a clinical diagnosis must not be given with radiological findings alone.

Legends

Fig 1. NCCT neck Sagittal reconstruction with bone window setting shows multiple block vertebrae throughout lower cervical spine (C5- T1). The facet joints are fused. Fused bodies appear hypoplastic, with reduced AP diameter and anterior concavity in vertebral bodies giving wasp waist appearance (red arrow).

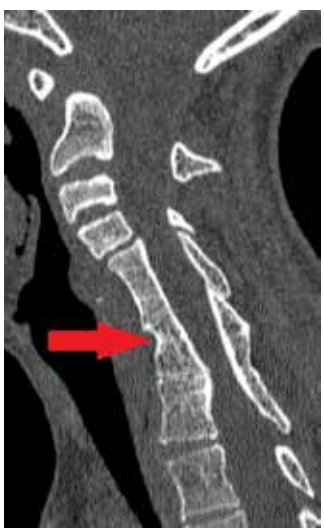
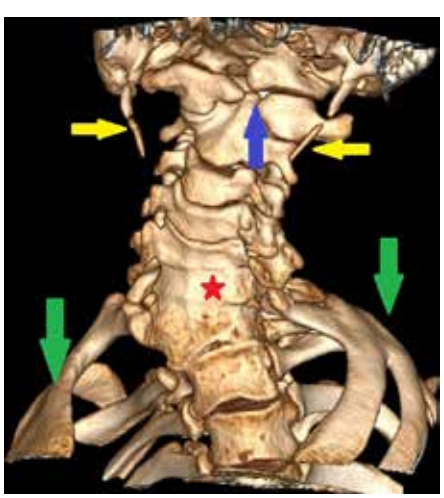


Fig 2. NCCT neck with volume rendering with 3D reconstruction shows a dextroconvex cervical scoliosis , butterfly vertebra (C1) (blue arrow), multiple block vertebrae(C5- T1) (red asterisk), hemivertebra (C7) and bilateral fork ribs (green arrows). Calcified bilateral stylohyoid ligaments seen (yellow arrows).



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